

## THE HUMAN GENOME PROJECT\*

LEON E. ROSENBERG, M.D.

Dean

Yale University School of Medicine  
New Haven, Connecticut

WE ARE HERE TODAY TO discuss the widely heralded human genome project, by all accounts a bold venture whose aim is to map and ultimately to sequence all of humankind's genetic material, arranged as it is in 22 different kinds of autosome chromosomes and two different kinds of sex chromosomes. I need not tell you that a great deal has been said and written about this project, some wise and necessary, some unwise, unnecessary and even foolish, as befits today's date, April 1, 1991. This project has been heralded as the biomedical equivalent of landing a man on the moon or building a space station. It has been likened to climbing Mount Everest; it has been called the definitive "book of man." It has also been decried as a Pandora's box which, when opened, will release a multitude of noxious, perhaps even deadly, vapors on our society, some of these vapors being medical, others social, legal, and ethical.

From my vantage point as a physician and a scientist who has spent his entire academic career in the field of human genetics, I cannot help but be a bit bemused by all this "hype." You see, the discipline of genetics has been attracting this kind of attention and this kind of exaggeration for much of this century, but most prominently since 1953, when Watson and Crick's Double Helix paper in *Nature* changed irrevocably the course of modern biology and medicine. I suspect many of you know, though it may bear repeating for the younger members of this audience, that every significant application of genetics toward humankind and human disease has been met with enormous interest and with equally enormous controversy.

In the 1960s the issue was newborn screening for genetic disease. To clinical geneticists like me, such screening was a means of early diagnosis, to be followed by effective treatment aimed at preventing serious consequences as in screening newborns for phenylketonuria and putting them on a low phenylalanine diet in order to prevent the mental retardation which is the

---

\*Presented as part of a *Conference on The Human Genome Project: An Agenda for Science and Society* held by the New York Academy of Medicine April 1, 1991.

critical and regular outcome of the untreated disorder. But such newborn screening raised for some the issue of stigmatization, discrimination, and prejudice. For others, consequences as dire as genocide were forecast.

In the 1970s the issue was prenatal diagnosis. To the clinical geneticist this was a means of detecting dread disorders such as anencephally, trisomy 18 or Tay-Sachs Disease in utero, thereby allowing those families who wished to avail themselves of this information to terminate pregnancies for cause. You all know that this matter has become embroiled in the still searing societal debate about abortion which rages to this very day.

In the 1980s the issue was somatic cell and germ line gene therapy. To the clinical geneticist, such treatment was a means of curing such diseases as beta thalassemia, sickle cell anemia, and severe combined immunodeficiency by transferring normal genes into cells of patients containing specific mutant ones. Once again, there were those who rose to decry this therapeutic modality as a fundamental danger to the way homo sapiens behaves and procreates.

It is, therefore, from this personal professional perspective that I have seen the genome project as another significant step in understanding human variation and its implications for health and disease. I have never seen it as an ultimate anything, just another step, another positive step in humankind's understanding of himself and his world. It was that perspective that led me, as a member of the National Research Council Committee which recommended embarking upon the human genome project, to support the consensus position of that group. I recognized, of course, that this was an expensive project and that it was a long-range project. But I never saw it as the dramatic issue which it seems to continue to be in the minds of scientists and the lay public. I would posit, and we shall see this as the day goes on, that much of the controversy about the genome project relates to the climate in which science and scientific funding exist today rather than to the project itself. I have a sneaking suspicion that, as the day goes on, you will hear rather different perspectives from the one I have just articulated.